SP110 gene

SP110 nuclear body protein

Normal Function

The *SP110* gene provides instructions for making a protein called SP110 nuclear body protein, which is a component of cellular structures called nuclear bodies. Nuclear bodies are located within the nuclei of cells, where they help control the activity of certain genes. Nuclear bodies are also involved in the regulation of cell division, the self-destruction of cells that are damaged or no longer needed (apoptosis), and the normal function of the immune system.

SP110 nuclear body protein is active primarily in immune system cells called leukocytes and in the spleen. It likely helps regulate the activity of genes needed for the body's immune response to foreign invaders (such as viruses and bacteria).

Health Conditions Related to Genetic Changes

hepatic veno-occlusive disease with immunodeficiency

At least five mutations in the *SP110* gene have been found to cause hepatic veno-occlusive disease with immunodeficiency (VODI). Each of these mutations leads to the production of a nonfunctional version of SP110 nuclear body protein. A lack of functional protein impairs the immune system's ability to fight off foreign invaders, allowing recurrent and persistent infections to develop. It is unclear how the loss of SP110 nuclear body protein disrupts blood flow in the liver, leading to enlargement of the liver (hepatomegaly), a buildup of scar tissue (hepatic fibrosis), and liver failure.

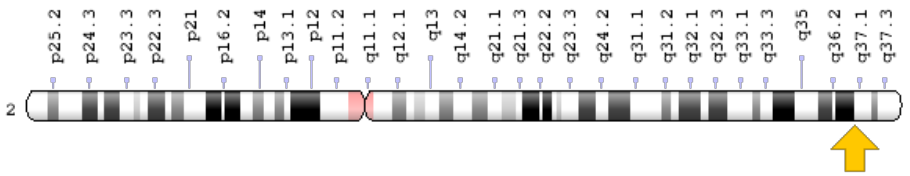
other disorders

Several common variations (polymorphisms) in the *SP110* gene have been studied as risk factors for lung (pulmonary) tuberculosis. This disease is a contagious bacterial infection. At least one study has found that certain variations in the *SP110* gene may influence the risk of infection with the bacteria that cause pulmonary tuberculosis. However, several other studies have not found such an association. *SP110* variations are probably not a major genetic risk factor for this disease.

Chromosomal Location

Cytogenetic Location: 2q37.1, which is the long (q) arm of chromosome 2 at position 37.1

Molecular Location: base pairs 230,165,466 to 230,225,729 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- FLJ22835
- IFI41
- IFI75
- interferon-induced protein 41, 30kD
- interferon-induced protein 41/75
- interferon-induced protein 75, 52kD
- IPR1
- nuclear body protein SP110
- phosphoprotein 41
- phosphoprotein 75
- SP110_HUMAN
- speckeled, 110-KD
- transcriptional coactivator Sp110

Additional Information & Resources

Educational Resources

- Nuclear Protein Database: PML Nuclear Bodies
  http://npd.hgu.mrc.ac.uk/user/compartment?page=pml

GeneReviews

- Hepatic Veno-Occlusive Disease with Immunodeficiency
  https://www.ncbi.nlm.nih.gov/books/NBK1271

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SP110%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- NUCLEAR BODY PROTEIN SP110
  http://omim.org/entry/604457

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SP110%5Bgene%5D
- HGNC Gene Family: Minor histocompatibility antigens
  http://www.genenames.org/cgi-bin/genefamilies/set/870
- HGNC Gene Family: PHD finger proteins
  http://www.genenames.org/cgi-bin/genefamilies/set/88
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=5401
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3431
- UniProt
  http://www.uniprot.org/uniprot/Q9HB58

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